Carol Dobson-Stone

List of Publications by Year in descending order

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80 papers 4,984 citations

38 h-index 95266 68 g-index

87 all docs

87 docs citations

87 times ranked

7483 citing authors

| # | Article | IF | CITATIONS |
|----|---|--------------|-----------|
| 1 | A conserved sorting-associated protein is mutant in chorea-acanthocytosis. Nature Genetics, 2001, 28, 119-120. | 21.4 | 357 |
| 2 | Frontotemporal dementia and its subtypes: a genome-wide association study. Lancet Neurology, The, 2014, 13, 686-699. | 10.2 | 302 |
| 3 | McLeod neuroacanthocytosis: Genotype and phenotype. Annals of Neurology, 2001, 50, 755-764. | 5 . 3 | 244 |
| 4 | Analysis of the human VPS13 gene family. Genomics, 2004, 84, 536-549. | 2.9 | 190 |
| 5 | Chorein detection for the diagnosis of choreaâ€acanthocytosis. Annals of Neurology, 2004, 56, 299-302. | 5. 3 | 186 |
| 6 | CCNF mutations in amyotrophic lateral sclerosis and frontotemporal dementia. Nature Communications, 2016, 7, 11253. | 12.8 | 174 |
| 7 | Sigma nonopioid intracellular receptor 1 mutations cause frontotemporal lobar degeneration–motor neuron disease. Annals of Neurology, 2010, 68, 639-649. | 5. 3 | 168 |
| 8 | Methylation of the oxytocin receptor gene and oxytocin blood levels in the development of psychopathy. Development and Psychopathology, 2014, 26, 33-40. | 2.3 | 163 |
| 9 | Neuroinflammation in frontotemporal dementia. Nature Reviews Neurology, 2019, 15, 540-555. | 10.1 | 159 |
| 10 | The Brain-Derived Neurotrophic Factor Val66Met Polymorphism Predicts Response to Exposure Therapy in Posttraumatic Stress Disorder. Biological Psychiatry, 2013, 73, 1059-1063. | 1.3 | 139 |
| 11 | TMEM106B is a genetic modifier of frontotemporal lobar degeneration with C9orf72 hexanucleotide repeat expansions. Acta Neuropathologica, 2014, 127, 407-418. | 7.7 | 123 |
| 12 | Disturbances in selective information processing associated with the BDNF Val66Met polymorphism: Evidence from cognition, the P300 and fronto-hippocampal systems. Biological Psychology, 2009, 80, 176-188. | 2,2 | 117 |
| 13 | <i>C9ORF72</i> repeat expansion in clinical and neuropathologic frontotemporal dementia cohorts. Neurology, 2012, 79, 995-1001. | 1.1 | 108 |
| 14 | Polymorphisms in the oxytocin receptor gene are associated with the development of psychopathy. Development and Psychopathology, 2014, 26, 21-31. | 2.3 | 105 |
| 15 | Preliminary Evidence of the Short Allele of the Serotonin Transporter Gene Predicting Poor Response to Cognitive Behavior Therapy in Posttraumatic Stress Disorder. Biological Psychiatry, 2010, 67, 1217-1219. | 1.3 | 98 |
| 16 | COMT Val108/158Met polymorphism effects on emotional brain function and negativity bias. Neurolmage, 2010, 53, 918-925. | 4.2 | 98 |
| 17 | Comparison of fluorescent single-strand conformation polymorphism analysis and denaturing high-performance liquid chromatography for detection of EXT1 and EXT2 mutations in hereditary multiple exostoses. European Journal of Human Genetics, 2000, 8, 24-32. | 2.8 | 85 |
| 18 | An Exploration of the Serotonin System in Antisocial Boys with High Levels of Callous-Unemotional Traits. PLoS ONE, 2013, 8, e56619. | 2.5 | 83 |

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|----|---|------|-----------|
| 19 | Reduced glucocerebrosidase activity in monocytes from patients with Parkinson's disease. Scientific Reports, 2018, 8, 15446. | 3.3 | 82 |
| 20 | Hailey-Hailey Disease: Molecular and Clinical Characterization of Novel Mutations in the ATP2C1 Gene. Journal of Investigative Dermatology, 2002, 118, 338-343. | 0.7 | 81 |
| 21 | Cerebellar Integrity in the Amyotrophic Lateral Sclerosis - Frontotemporal Dementia Continuum. PLoS ONE, 2014, 9, e105632. | 2.5 | 79 |
| 22 | DNA methylation of the <i>MAPT</i> gene in Parkinson's disease cohorts and modulation by vitamin E <i>In Vitro</i> . Movement Disorders, 2014, 29, 1606-1614. | 3.9 | 79 |
| 23 | Brain derived neurotrophic factor Val66Met polymorphism, the five factor model of personality and hippocampal volume: Implications for depressive illness. Human Brain Mapping, 2009, 30, 1246-1256. | 3.6 | 78 |
| 24 | Variation in the oxytocin receptor gene is associated with increased risk for anxiety, stress and depression in individuals with a history of exposure to early life stress. Journal of Psychiatric Research, 2014, 59, 93-100. | 3.1 | 78 |
| 25 | A Polymorphism of the MAOA Gene is Associated with Emotional Brain Markers and Personality Traits on an Antisocial Index. Neuropsychopharmacology, 2009, 34, 1797-1809. | 5.4 | 74 |
| 26 | Pedigree with frontotemporal lobar degeneration $\hat{a}\in$ motor neuron disease and Tar DNA binding protein-43 positive neuropathology: genetic linkage to chromosome 9. BMC Neurology, 2008, 8, 32. | 1.8 | 71 |
| 27 | Impact of the HTR3A gene with early life trauma on emotional brain networks and depressed mood. Depression and Anxiety, 2010, 27, 752-759. | 4.1 | 69 |
| 28 | Role of the Long Non-Coding RNA MAPT-AS1 in Regulation of Microtubule Associated Protein Tau (MAPT) Expression in Parkinson's Disease. PLoS ONE, 2016, 11, e0157924. | 2.5 | 68 |
| 29 | Comprehensive genetic diagnosis of tandem repeat expansion disorders with programmable targeted nanopore sequencing. Science Advances, 2022, 8, eabm5386. | 10.3 | 68 |
| 30 | Association between BDNF Val66Met polymorphism and trait depression is mediated via resting EEG alpha band activity. Biological Psychology, 2008, 79, 275-284. | 2.2 | 67 |
| 31 | Familial Temporal Lobe Epilepsy as a Presenting Feature of Choreoacanthocytosis. Epilepsia, 2005, 46, 1256-1263. | 5.1 | 62 |
| 32 | CYLD is a causative gene for frontotemporal dementia – amyotrophic lateral sclerosis. Brain, 2020, 143, 783-799. | 7.6 | 62 |
| 33 | Early Clinical Heterogeneity in Choreoacanthocytosis. Archives of Neurology, 2005, 62, 611. | 4.5 | 61 |
| 34 | C9ORF72 Repeat Expansion in Australian and Spanish Frontotemporal Dementia Patients. PLoS ONE, 2013, 8, e56899. | 2.5 | 56 |
| 35 | THE INTEGRATE MODEL OF EMOTION, THINKING AND SELF REGULATION: AN APPLICATION TO THE "PARADOX OF AGING". Journal of Integrative Neuroscience, 2008, 07, 367-404. | 1.7 | 48 |
| 36 | Genetic Polymorphisms in Sigma-1 Receptor and Apolipoprotein E Interact to Influence the Severity of Alzheimers Disease. Current Alzheimer Research, 2011, 8, 765-770. | 1.4 | 48 |

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|----|---|-----|-----------|
| 37 | Association of Variants in the <i>SPTLC1</i> Gene With Juvenile Amyotrophic Lateral Sclerosis. JAMA Neurology, 2021, 78, 1236. | 9.0 | 46 |
| 38 | Developments in neuroacanthocytosis: Expanding the spectrum of choreatic syndromes. Movement Disorders, 2006, 21, 1794-1805. | 3.9 | 44 |
| 39 | A Functional Polymorphism of the <i>MAOA</i> Gene Is Associated with Neural Responses to Induced Anger Control. Journal of Cognitive Neuroscience, 2014, 26, 1418-1427. | 2.3 | 44 |
| 40 | A C6orf10/LOC101929163 locus is associated with age of onset in C9orf72 carriers. Brain, 2018, 141, 2895-2907. | 7.6 | 39 |
| 41 | A Role for Transcription Factor GTF2IRD2 in Executive Function in Williams-Beuren Syndrome. PLoS ONE, 2012, 7, e47457. | 2.5 | 37 |
| 42 | Associations between the COMT Val/Metpolymorphism, early life stress, and personality among healthy adults. Neuropsychiatric Disease and Treatment, 2006, 2, 219-225. | 2.2 | 37 |
| 43 | Identification of a VPS13A founder mutation in French Canadian families with chorea-acanthocytosis. Neurogenetics, 2005, 6, 151-158. | 1.4 | 36 |
| 44 | Neural substrates of episodic memory dysfunction in behavioural variant frontotemporal dementia with and without C9ORF72 expansions. NeuroImage: Clinical, 2013, 2, 836-843. | 2.7 | 35 |
| 45 | A GENOTYPE-ENDOPHENOTYPE-PHENOTYPE PATH MODEL OF DEPRESSED MOOD: INTEGRATING COGNITIVE AND EMOTIONAL MARKERS. Journal of Integrative Neuroscience, 2007, 06, 75-104. | 1.7 | 33 |
| 46 | The association between the oxytocin receptor gene (OXTR) and hypnotizability. Psychoneuroendocrinology, 2013, 38, 1979-1984. | 2.7 | 32 |
| 47 | The contribution of BDNF and 5-HTT polymorphisms and early life stress to the heterogeneity of major depressive disorder: A preliminary study. Australian and New Zealand Journal of Psychiatry, 2012, 46, 55-63. | 2.3 | 30 |
| 48 | The underacknowledged PPA-ALS. Neurology, 2019, 92, e1354-e1366. | 1.1 | 29 |
| 49 | Serotonin 1B Receptor Gene (HTR1B) Methylation as a Risk Factor for Callous-Unemotional Traits in Antisocial Boys. PLoS ONE, 2015, 10, e0126903. | 2.5 | 28 |
| 50 | Distinct TDP-43 inclusion morphologies in frontotemporal lobar degeneration with and without amyotrophic lateral sclerosis. Acta Neuropathologica Communications, 2017, 5, 76. | 5.2 | 27 |
| 51 | Genomic Organization of the Human Gα14 and Gαq Genes and Mutation Analysis in Chorea–Acanthocytosis (CHAC). Genomics, 1999, 57, 84-93. | 2.9 | 25 |
| 52 | INTEGRATING OBJECTIVE GENE-BRAIN-BEHAVIOR MARKERS OF PSYCHIATRIC DISORDERS. Journal of Integrative Neuroscience, 2007, 06, 1-34. | 1.7 | 24 |
| 53 | Frontotemporal dementia–amyotrophic lateral sclerosis syndrome locus on chromosome 16p12.1–q12.2: genetic, clinical and neuropathological analysis. Acta Neuropathologica, 2013, 125, 523-533. | 7.7 | 24 |
| 54 | Clinical and Biological Correlates of White Matter Hyperintensities in Patients With Behavioral-Variant Frontotemporal Dementia and Alzheimer Disease. Neurology, 2021, 96, e1743-e1754. | 1.1 | 24 |

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| 55 | Chorea-acanthocytosis: Clinical and genetic findings in three families from the Arabian peninsula. Movement Disorders, 2003, 18, 403-407. | 3.9 | 22 |
| 56 | The functional epistasis of 5―HTTLPR and BDNF Val66Met on emotion processing: a preliminary study. Brain and Behavior, 2012, 2, 778-788. | 2.2 | 21 |
| 57 | Effect of stress gene-by-environment interactions on hippocampal volumes and cortisol secretion in adolescent girls. Australian and New Zealand Journal of Psychiatry, 2019, 53, 316-325. | 2.3 | 20 |
| 58 | Impact of 5-HTTLPR on SSRI serotonin transporter blockade during emotion regulation: A preliminary fMRI study. Journal of Affective Disorders, 2016, 196, 11-19. | 4.1 | 19 |
| 59 | Predicting Development of Amyotrophic Lateral Sclerosis in Frontotemporal Dementia. Journal of Alzheimer's Disease, 2017, 58, 163-170. | 2.6 | 17 |
| 60 | Interactions of OXTR rs53576 and emotional trauma on hippocampal volumes and perceived social support in adolescent girls. Psychoneuroendocrinology, 2020, 115, 104635. | 2.7 | 17 |
| 61 | IsCHCHD10Pro34Ser pathogenic for frontotemporal dementia and amyotrophic lateral sclerosis?: Figure 1. Brain, 2015, 138, e385-e385. | 7.6 | 16 |
| 62 | The impact of 5-HTTLPR on acute serotonin transporter blockade by escitalopram on emotion processing: Preliminary findings from a randomised, crossover fMRI study. Australian and New Zealand Journal of Psychiatry, 2014, 48, 1115-1125. | 2.3 | 14 |
| 63 | Effect of Fluvoxamine on Amyloid- \hat{l}^2 Peptide Generation and Memory. Journal of Alzheimer's Disease, 2018, 62, 1777-1787. | 2.6 | 12 |
| 64 | Association between serotonin transporter promoter polymorphisms and psychological distress in a diabetic population. Psychiatry Research, 2012, 200, 343-348. | 3.3 | 10 |
| 65 | Endogenous progesterone levels and frontotemporal dementia: modulation of TDP-43 and Tau levels in vitro and treatment of the A315T TARDBP mouse model. DMM Disease Models and Mechanisms, 2013, 6, 1198-204. | 2.4 | 10 |
| 66 | TDP-43 in the hypoglossal nucleus identifies amyotrophic lateral sclerosis in behavioral variant frontotemporal dementia. Journal of the Neurological Sciences, 2016, 366, 197-201. | 0.6 | 10 |
| 67 | Gene Expression Imputation Across Multiple Tissue Types Provides Insight Into the Genetic Architecture of Frontotemporal Dementia and Its Clinical Subtypes. Biological Psychiatry, 2021, 89, 825-835. | 1.3 | 10 |
| 68 | Choreoacanthocytosis in a Mexican Family. Archives of Neurology, 2007, 64, 1661. | 4.5 | 9 |
| 69 | Effect of PSEN1 mutations on MAPT methylation in early-onset Alzheimer's disease. Current Alzheimer Research, 2015, 12, 745-751. | 1.4 | 9 |
| 70 | GSK3B and MAPT Polymorphisms Are Associated with Grey Matter and Intracranial Volume in Healthy Individuals. PLoS ONE, 2013, 8, e71750. | 2.5 | 8 |
| 71 | Finding MAPT Mutations in Frontotemporal Dementia and Other Tauopathies. Methods in Molecular Biology, 2017, 1523, 307-324. | 0.9 | 6 |
| 72 | The complex relationship between genotype, pathology and phenotype in familial dementia. Neurobiology of Disease, 2020, 145, 105082. | 4.4 | 6 |

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| 73 | Reply: CYLD variants in frontotemporal dementia associated with severe memory impairment in a Portuguese cohort. Brain, 2020, 143, e68-e68. | 7.6 | 4 |
| 74 | Mendelian randomization implies no direct causal association between leukocyte telomere length and amyotrophic lateral sclerosis. Scientific Reports, 2020, 10, 12184. | 3.3 | 4 |
| 75 | Examining the presence and nature of delusions in Alzheimer's disease and frontotemporal dementia syndromes. International Journal of Geriatric Psychiatry, 2022, 37, . | 2.7 | 4 |
| 76 | P1-039: MAPT METHYLATION IN ALZHEIMER'S DISEASE. , 2014, 10, P317-P318. | | 3 |
| 77 | Rapid in vitro quantification of TDP-43 and FUS mislocalisation for screening of gene variants implicated in frontotemporal dementia and amyotrophic lateral sclerosis. Scientific Reports, 2021, 11, 14881. | 3.3 | 3 |
| 78 | The treA gene of Bacillus subtilis is a suitable reporter gene for the archaeon Methanococcus voltae. FEMS Microbiology Letters, 1998, 164, 237-242. | 1.8 | 2 |
| 79 | Cerebellar integrity and contributions to cognition in C9orf72-mediated frontotemporal dementia. Cortex, 2022, 149, 73-84. | 2.4 | 2 |
| 80 | Schizotypal traits across the amyotrophic lateral sclerosis–frontotemporal dementia spectrum: pathomechanistic insights. Journal of Neurology, 2022, , 1. | 3.6 | O |